

## Chapter 1: Human Genome

### 1.1. INTRODUCING GENES AND GENOME

- **Genetics** – study of heredity, traits and their variations.
  - study of how traits are transmitted.
- **Heredity** – transmission of traits and biological information.
- **Genes** – basic unit of heredity.
  - instruct cell's production of proteins.
  - consist of long DNA.
  - provide-encoding genes are scattered among 3.2 billion DNA bases in each set of chromosomes.
- **Phenotype** – control the characteristics of every individual.
- **DNA (Deoxyribonucleic Acid)** – transmits information in sequence of nitrogenous bases.
- **Genome** – make up of complete set of genetic instructions characteristic of each an organism.
  - all of our cells contain two sets copies of genome.
- **Genomics** – scientific field that analyzes genomes.
  - reveals how closely related are different species.

### 1.2 LEVELS OF GENETICS AND GENOMICS

- **DNA (Deoxyribonucleic Acid)**
  - Shape: double helix
  - Consist of: alternating sugar and phosphates
  - Function: provide information to manufacture specific proteins



- 4 types of nitrogenous bases:
  - Adenine (A) and Thymine (T)
  - Cytosine (C) and Guanine (G)
  - Each set of these complementary bases is a code for a particular amino acid.
- **Amino Acid** – building blocks of protein.
  - align and link like snap beads forming protein

### 1.3. THREE PROCESSES OF ACCESSING GENETIC INFORMATION

- **DNA replication** – (1) chains of double helix untwist and separate.
  - (2) each full helix is a new partner chain from two DNA bases.
- **Transcription** – (3) copies the sequence of one strand of DNA molecule into mRNA.
- **Translation** – (4) each 3 RNA bases is a new attract another type of RNA, that functions as a connector, bringing in a particular amino acid.
  - (5) Amino acids align and link to form protein.

### 1.2 THE HUMAN GENOME

- **20,125 protein-encoding genes** comprise the genome.
  - Accounts: 1.5 percent of human genome, 8.5 percent of known genetic diseases.
- **Alloles** ("gene variants") – some proteins – encoding genes may vary in DNA base sequence from person to person that is passed to the next generation.
- **Mutations** ("changed gene") – variation or change in genes, causing a disease.

### 1.3. INHERITED DISEASES

- **Cystic Fibrosis** – inherited disease
  - Change of a "C" to "T" inserts amino acid rather than the amino acid glycine which results to proteins becoming abnormal
  - Cystic Fibrosis Transmembrane

# Biology Chapter 1the Human Genome

**Joacim Rocklöv**



## **Biology Chapter 1the Human Genome:**

Handbook of Systems Biology Marian Walhout, Marc Vidal, Job Dekker, 2012-12-31 This book provides an entry point into Systems Biology for researchers in genetics molecular biology cell biology microbiology and biomedical science to understand the key concepts to expanding their work Chapters organized around broader themes of Organelles and Organisms Systems Properties of Biological Processes Cellular Networks and Systems Biology and Disease discuss the development of concepts the current applications and the future prospects Emphasis is placed on concepts and insights into the multi disciplinary nature of the field as well as the importance of systems biology in human biological research Technology being an extremely important aspect of scientific progress overall and in the creation of new fields in particular is discussed in boxes within each chapter to relate to appropriate topics 2013 Honorable Mention for Single Volume Reference in Science from the Association of American Publishers PROSE Awards Emphasizes the interdisciplinary nature of systems biology with contributions from leaders in a variety of disciplines Includes the latest research developments in human and animal models to assist with translational research Presents biological and computational aspects of the science side by side to facilitate collaboration between computational and biological researchers **Genes and Evolution**

, 2016-06-06 Genes and Evolution the latest volume in the Current Topics in Developmental Biology series covers genes and evolution with contributions from an international board of authors The chapters provide a comprehensive set of reviews covering such topics as genes and plant domestication gene networks phenotypic loss in vertebrates reproducible evolutionary changes and epithelial tissue Covers the area of genes and evolution Contains invaluable contributions from an international board of authors Provides a comprehensive set of reviews covering such topics as genes and plant domestication gene networks phenotypic loss in vertebrates reproducible evolutionary changes and epithelial tissue

*Introduction to Genomics* Arthur M. Lesk, 2017 The most up to date and complete textbook for first time genomics students Introduction to Genomics offers a fascinating insight into how organisms differ or match how different organisms evolved how the genome is constructed and how it operates and what our understanding of genomics means in terms of our future health and wellbeing This fully updated and restructured new edition which includes two new chapters takes account of new developments and technologies presenting a logical and coherent overview of genome science today The author s widely praised writing style leads the reader through a conceptually challenging subject in a clear lucid way building confidence in and enthusiasm for the subject at the outset Broad and fascinating range of real world examples which are also relevant across genet **Aging** Carsten Carlberg, Stine M. Ulven, Eunike Velleuer, 2024-07-22 Aging is a topic that concerns all of us since none of us can escape it The molecular and cellular process is built in every of the billions of cells forming our body Some of these cells such as immune cells and red blood cells live only for a few days to weeks and get life long constantly replaced by cells produced in the bone marrow In contrast there are cells such as neurons and memory

lymphocytes that get as old as we get The process of aging limits our maximal life span which is for us humans 120 years However only a very few individuals reached this age How did their life differ from others that died decades earlier Is it just the absence of life threatening disease paired with a more healthy life style Or is it build in in our genome or epigenome In this book we try to give answers to these questions from the perspectives of evolution our genome the epigenomes of our different tissues and cell types and the functionality of our cells We should try to understand ourselves in detail as well as in a global setting Basic biology explains cellular mechanisms such as growth differentiation and cell death which make life as a whole possible Every human organism represents a complex interplay between hundreds of different cell types forming distinctive tissues and organs with specialized tasks These processes need to be highly orchestrated especially during development maintenance and aging Studying the cellular and molecular basis of aging is one of the most fascinating areas but also a great challenge Nevertheless research made the biggest steps in elucidating biological processes via studying malfunctions of normal mechanisms leading to different diseases such as progeroid syndrome and cancer We will start this book with the understanding of the human genome in relation to principles of evolution Then we will explain the basics of gene regulation and epigenetics i e the interplay of transcription factors and chromatin Next we will shift to cellular mechanisms of aging and discuss then the impact of nutrition and immunity on the aging process In the following the relation of aging to so called aging related common diseases such as type 2 diabetes atherosclerosis cancer and Alzheimer Do we get these diseases because we are aging or are we aging because we get one of these diseases The book will end how we can slow down the aging process so that we can age healthy In short healthy aging is not an option but is a must An ancient poem says Teach us to number our days that we may get a heart of wisdom It is up to each one of us and a daily decision to live a healthy lifestyle and to be aware of the unique gift of live we all have

*Core Laboratory Technologies in Clinical Immunology E-Book* Robert R. Rich, 2018-10-24 Edited by clinical immunology expert Dr Robert R Rich this concise focused title covers today s most important technologies used in the diagnosis and evaluation of immunologic disease Core Laboratory Technologies in Clinical Immunology is ideal for immunology researchers and scientists as well as immunologists and others interested in the principles and uses of current lab technologies in immunology Focuses on how today s technologies relate to the diagnosis of disease including state of the art technologies that are significantly impacting cancer therapy research Covers flow cytometry assessment of functional immune responses in lymphocytes assessment of neutrophil function molecular methods and more Provides information of special interest to researchers and scientists who are directly involved in the rapidly changing world of clinical immunology as well as immunologists oncologists and medical technology and biomedical engineers Consolidates today s available information and guidance into a single convenient resource

*Biology* Neil A. Campbell, Jane B. Reece, 2005 Neil Campbell and Jane Reece s BIOLOGY remains unsurpassed as the most successful majors biology textbook in the world This text has invited more than 4 million students into the study of this

dynamic and essential discipline The authors have restructured each chapter around a conceptual framework of five or six big ideas An Overview draws students in and sets the stage for the rest of the chapter each numbered Concept Head announces the beginning of a new concept and Concept Check questions at the end of each chapter encourage students to assess their mastery of a given concept New Inquiry Figures focus students on the experimental process and new Research Method Figures illustrate important techniques in biology Each chapter ends with a Scientific Inquiry Question that asks students to apply scientific investigation skills to the content of the chapter

**Creasy and Resnik's Maternal-Fetal Medicine: Principles and Practice E-Book** Robert Resnik, Robert K. Creasy, Jay D. Iams, Charles J. Lockwood, Thomas Moore, Michael F Greene, 2008-11-25 In your practice you require advanced knowledge of the obstetrical medical genetic and surgical complications of pregnancy and their effects on the mother and fetus With both basic science and clinical information six new chapters and an updated color design you need look no further than the 6th edition of this long time best seller Includes both basic science and clinical information to give you comprehensive knowledge of the biology of pregnancy Acts as an excellent resource for OB GYNs studying for their Maternal Fetal Medicine boards and for practitioners who need quick access to practical information Provides an updated and focused reference list to keep you up to date on the standards of care in maternal fetal medicine today Keeps you current with a new section Disorders at the Maternal Fetal Interface and 6 new chapters Biology of Parturition Developmental Origins of Health and Disease Intrapartum Assessment of Fetal Health Pathogenesis of Pre term Birth Maternal and Fetal Infectious Disorders and Benign Gynecological Conditions of Pregnancy Features over 50% new authorship with increased focus on international perspectives Includes the following hot topics in Maternal Fetal Medicine o Biology of Parturition o Fetal Growth o Prenatal Genetic Screening and Diagnosis o Fetal Cardiac Malformations and Arrhythmias o Thyroid Disease and Pregnancy o Management of Depression and Psychoses during Pregnancy and the Puerperium Focuses on evidence based medicine the current best practice in MFM for diagnosing and treating high risk pregnancies Includes new illustrations and an updated color design

**What is Human** Terri Lynn Peterson, 2000

**Loose-leaf Version for Molecular Cell Biology** Harvey Lodish, Arnold Berk, Chris A. Kaiser, Monty Krieger, Anthony Bretscher, Hidde Ploegh, Angelika Amon, Kelsey C. Martin, 2016-04-01 With its acclaimed author team cutting edge content emphasis on medical relevance and coverage based on key experiments Molecular Cell Biology has justly earned an impeccable reputation as an exciting and authoritative text Avoiding an encyclopedic approach the book grounds its coverage in the experiments that define our understanding of cell biology engaging students with the exciting breakthroughs that define the field s history and point to its future The authors all world class researchers and teachers incorporate medically relevant examples where appropriate to help illustrate the connections between cell biology and health and human disease

**Genetic Variation** Michael P. Weiner, Stacey B. Gabriel, J. Claiborne Stephens, 2007 This is the first compendium of protocols specifically geared towards genetic variation studies It includes detailed step by step experimental

protocols that cover the complete spectrum of genetic variation in humans and model organisms along with advice on study design and analyzing data     *API Textbook of Medicine* Yash Pal Munjal,2012     *Genetics* Daniel L. Hartl,2009

Principles of Genetics D. Peter Snustad,Michael J. Simmons,2016-01-04 Principles of Genetics is one of the most popular texts in use for the introductory course It opens a window on the rapidly advancing science of genetics by showing exactly how genetics is done Throughout the authors incorporate a human emphasis and highlight the role of geneticists to keep students interested and motivated The seventh edition has been completely updated to reflect the latest developments in the field of genetics Principles of Genetics continues to educate today s students for tomorrows science by focusing on features that aid in content comprehension and application     *Principles of Life* David M. Hillis,David E. Sadava,Richard Hill,Mary V. Price,2014-07-15 With its first edition Principles of Life provided a textbook well aligned with the recommendations proposed in BIO 2010 Transforming Undergraduate Education for Future Research Biologists and Vision and Change in Undergraduate Biology Education Now Principles of Life returns in a thoroughly updated new edition that exemplifies the reform that is remaking the modern biology classroom     *Loose-leaf Version for Principles of Life* David M. Hillis,David E. Sadava,Richard W. Hill,Mary V. Price,2013-12-01 With its first edition Principles of Life provided a textbook well aligned with the recommendations proposed in BIO 2010 Transforming Undergraduate Education for Future Research Biologists and Vision and Change in Undergraduate Biology Education Now Principles of Life returns in a thoroughly updated new edition that exemplifies the reform that is remaking the modern biology classroom     **Harper's Illustrated Biochemistry, Thirty-Second Edition** Peter J. Kennelly,Kathleen M. Botham,Owen McGuinness,Victor W. Rodwell,P. Anthony Weil,2022-08-30 Gain a thorough understanding of the principles of biochemistry as they relate to clinical medicine A Doody s Core Title for 2024 2023 The Thirty Second Edition of Harper s Illustrated Biochemistry combines top quality full color illustrations with authoritative integrated coverage of biochemical disease and clinical information Featuring numerous medically relevant examples this respected text presents a clear succinct review of the fundamentals that every student must understand in order to succeed in medical school All 58 chapters help you understand the medical relevance of biochemistry Full color presentation with 600 illustrations Chapters have been updated to reflect the latest information Case studies emphasize the clinical relevance of biochemistry Review questions follow each of the 11 sections Boxed objectives define the goals of each chapter Tables encapsulate important information Each chapter contains a section on biomedical importance and a summary of the topics covered Applauded by medical students for its current and engaging style Harper s Illustrated Biochemistry is an essential for USMLE review and the single best reference for learning the clinical relevance of any biochemistry topic     Harper's Illustrated Biochemistry Thirty-First Edition Victor W. Rodwell,David Bender,Kathleen M. Botham,Peter J. Kennelly,P. Anthony Weil,2018-06-22 Gain a full understanding of the principles of biochemistry as it relates to clinical medicine A Doody s Core Title for 2021 The Thirty First Edition of Harper s Illustrated Biochemistry continues to

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Thompson,2013-11-22 A complete introductory text on how to integrate basic genetic principles into the practice of clinical medicine Medical Genetics is the first text to focus on the everyday application of genetic assessment and its diagnostic therapeutic and preventive implications in clinical practice It is intended to be a text that you can use throughout medical school and refer back to when questions arise during residency and eventually practice Medical Genetics is written as a narrative where each chapter builds upon the foundation laid by previous ones Chapters can also be used as stand alone learning aids for specific topics Taken as a whole this timely book delivers a complete overview of genetics in medicine You will find in depth expert coverage of such key topics as The structure and function of genes Cytogenetics Mendelian inheritance Mutations Genetic testing and screening Genetic therapies Disorders of organelles Key genetic diseases disorders and syndromes Each chapter of Medical Genetics is logically organized into three sections Background and Systems Includes the basic genetic principles needed to understand the medical application Medical Genetics Contains all the pertinent information necessary to build a strong knowledge base for being successful on every step of the USMLE Case Study Application Incorporates case study examples to illustrate how basic principles apply to real world patient care Today with every component of health care delivery requiring a working knowledge of core genetic principles Medical Genetics is a true must read for every clinician Basic Genetics Robert F. Weaver,Philip W. Hedrick,1995 This text provides a balanced coverage of clinical and molecular genetics Experimental highlights and extensive use of learning aids are used throughout After a broad introduction to the topic the book is divided into 3 parts Part one explores Mendelian genetics including

chromosomes and genetic linkage Part two looks at molecular genetics covering chemistry of a gene repluation and recombination of genes and transcription and its control in prokaryotes The final part introduces population genetics and discusses some of their extensions and applications      **The Heart, Arteries and Veins** John Willis Hurst,Robert C. Schlant,R. Wayne Alexander,1994



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